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2013

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Mia Alessandra: My Strength

Life With Juvenile Idiopathic Arthritis

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Report

Presented to the Faculty of the Graduate School of

the University of Texas at Austin

in Partial Fulfillment

of the Requirements

for the Degree of

Master of Arts

The University of Texas at Austin

May 2013

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The University of Texas at Austin, 2013

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Mia Alessandra Nieto is an 8-year old living with Juvenile Idiopathic Arthritis (JIA) in Austin, Texas. When she was diagnosed at 10 months, she was the youngest child ever diagnosed with JIA in the state of Texas. However, it took 37 days to confirm her condition because there is an immense lack on knowledge in the field of pediatric rheumatology among general practitioners despite the fact that JIA is the most prevalent chronic condition in children in the United States with over 300,000 diagnosed. This is an overview of Mia's story, along with information regarding the lack of knowledge on the condition not only in the general population but mainly and more importantly among the medical professionals in the United States.

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The name Mia Alessandra means “my strength” in Spanish, and this name suits little Mia Alessandra Nieto perfectly. At first glance, 8-year-old Mia seems like any other second grader; she is playing softball for the first time, learning to read short chapter books, loves to play on jungle gyms and to play with friends. Her infectious laughter, free spirit, and ability to connect and converse with anyone -- young or old, stranger or close friend -- make her an instant light in any room. From looking at her sweet face and sensing her bright and happy nature, it would be nearly impossible to fathom that Mia has had multiple MRIs (Magnetic Resonance Images) on nearly every single part of her body, 6 rounds of steroid joint injections, over 20 X-rays, a bunionectomy at the age of three and a half, and has been on multiple injectable chemotherapy drugs, NSAIDS (non-steroidal anti-inflammatory drugs), steroids, and biologic infusions for nearly her entire eight years of life.

On April 7th 2005, when Mia was just 9 months old, her parents Chris and Erika brought her to the hospital after she had significant gastrointestinal issues for over a week. When nothing serious was found, they went home with no major concerns. Then, on April 15th she awoke crying in the middle of the night with a fever of around 104 degrees and a swollen wrist. Her parents brought her to the pediatrician, but nothing was determined about her sickness. They went back to the pediatrician again after her fever went up to 105 degrees. The next 37 days were filled with hospital visits, appointments with their family pediatrician and appointments with infectious disease specialists where now 10 month old Mia was constantly poked and prodded. She was incessantly being examined, getting blood

drawn for a variety of tests, having stool samples done, being X-rayed (she even had x-rays taken of her spine) and bone scans with no conclusive results or diagnosis. Throughout this time, her persistent fever remained at around 104/105 degrees. Finally, after more than a month of this, their infectious disease specialist referred them to a pediatric rheumatologist in Houston, TX, two and a half hours from where the family lived outside of Austin, TX. When they were able to be seen by this doctor, Mia's condition was finally diagnosed: Systemic-Onset Juvenile Idiopathic Arthritis. At only nine months, Mia was the youngest child ever diagnosed with arthritis in the state of Texas.

Then began the treatment. For nearly all of her entire eight years of life, Mia has had drugs in her system in order to allow her to move freely, keep her inflammation and joint deterioration down, and keep her functioning as normally as possible. When she was first diagnosed, Mia was put on steroids and Naproxen, an NSAID. Throughout the years she has been on various drugs such as Humira, Enbrel, Methotrexate, Naproxen, and Actemra, in addition to steroids. Now, as an 8 year old, Mia has a weekly injection of Methotrexate, an hour and a half infusion every two weeks of the biologic medication Actemra, and a daily oral pill of Naproxen. Although she is able to run and play, Mia still experiences daily stiffness, soreness and pain. She also has no cartilage in her left ankle because it has all been destroyed by her immune system and she has uncontrollable swelling in her ankles and wrists. Her pediatric rheumatologist, Dr. Ruy Carrasco of Dell Children's

Medical Center in Austin, Texas told her parents at one point that Mia had “the worst arthritis [he had] ever seen.”

Although, for most, it may seem shocking to hear that a nine---month old was diagnosed with arthritis, Juvenile Idiopathic Arthritis, more commonly referred to as JIA, is actually the most prevalent chronic condition in children in the United States. According to a study done by the Centers for Disease Control and Prevention (CDC) in 2007, there were 294,000 diagnosed cases of arthritis or other rheumatic conditions in the US alone, and because rheumatic conditions can be difficult to diagnose, it is quite possible that this number is under---representative of the actual number of children living with arthritis or similar rheumatic conditions in the US. Even without speculation that this number is inaccurate, 294,000 cases means that 1 in every 250 children is affected by a rheumatic condition (this includes conditions other than arthritis). In comparison to better known chronic conditions, the World Health Organization (WHO) reports that Cystic Fibrosis has a prevalence rate of 1 in every 3,500 births, CDC reports in 2002 that Down Syndrome has a prevalence rate of 1 in every 1,000 births, and the CDC also reports that prevalence rates for juvenile diabetes are only 1.7 per 1,000.

Although these statistics show that rheumatic conditions, arthritis specifically, are the most prevalent childhood afflictions, they are generally unheard of in the general population. When most people hear about children with arthritis they are bewildered because arthritis is known in the United States as an “old---person’s disease.” Although adults do make up the vast majority of the 50 million

Americans diagnosed with arthritis that fact should not undermine the fact that it is the most prevalent chronic condition in children. Most people think of arthritis as causing pain and inflammation of a joint, but a more accurate definition, provided by the Arthritis Foundation, is that arthritis is “a complex family of musculoskeletal disorders consisting of more than 100 different diseases or conditions that can affect people of all ages, races and genders.” A musculoskeletal condition is one characterized by the involvement of the muscle and bones, and juvenile arthritis involves not only the bones and muscles but also all organs and systems of the body. According to *Pediatric Rheumatology in Clinical Practice* by Dr. Patricia Woo, Dr. Ronald Laxer and Dr. David Sherry, rheumatology as a whole is unique in that it’s diseases, arthritis specifically, “can affect a person wherever there is a blood vessel or where there isn't.”

Arthritis is well known as a medical condition that causes pain, swelling, and inflammation of the joints and, while that is accurate, that is not all there is to the disease. Arthritis is an autoimmune condition, meaning that it is caused by one’s own immune system. When an immune system is working properly, it attacks foreign bodies, usually viruses and bacteria and microorganisms that cause harm to our bodies. When a person has an autoimmune condition like arthritis, the immune system incorrectly thinks that parts of it’s own body are foreign and therefore need to be attacked. In arthritis specifically, the immune system does not recognize the joints as being from the same body and therefore safe. It sees the joints as a threat and attacks them, which can cause the joints to become inflamed, painful, swollen,

and/or stiff. It can also cause deterioration of the joint if left untreated. However, it is important to note that arthritis does not simply affect joints only; because it is the immune system that is causing the damage, it can do damage anywhere in the body. Although there is some evidence that points to arthritis being related to a genetic predisposition, there is no known cause. In fact, the word “idiopathic” means “of unknown origin.” Not only is there no known cause, there is no known cure.

JIA is a very broad umbrella term for juvenile arthritis. In reality, there are seven different, very distinct, kinds of Juvenile Idiopathic Arthritis. The first and most common type, which affects around 50---60% of children diagnosed with JIA, is Oligoarticular JIA. Persistent Oligoarticular JIA, which is characterized by the arthritis affecting four or fewer joints in the first six months after diagnosis is most common in girls ages 1---3 and they typically first feel pain and swelling in the knee or ankle. If, after six months, the arthritis begins to affect more than four joints, it then is known as Extended Oligoarticular JIA. Although this is the most common kind of juvenile arthritis, there is an interesting curveball, so to speak, with diagnosis because around 25% of children diagnosed do not report feeling pain. This type of arthritis is one where it is not irregular for parts of the body other than the joint can be affected. Muscle atrophy can occur due to chronic swelling and pain, as well as leg length discrepancies. There is also the problem of uveitis, which is inflammation of the eye. If this condition is present and is not caught by an ophthalmologist, chronic inflammation of the eye can cause serious vision problems and even blindness in children with JIA.

Psoriatic JIA is a form of arthritis that presents with psoriasis. This means that a child has symptoms not only of arthritis but also has either dactylitis, which is the inflammation of a digit such as a finger or toe, nail abnormalities, such as multiple dents or pits in fingernails or toenails, or even simply has a first-degree relative (a parent or sibling) with psoriasis. Children who are diagnosed with this type of JIA usually end up having longer-lasting symptoms than children with other types. Not only that but also “a small but significant percentage (up to 10%) may be disabled” according to *Pediatric Rheumatology in Clinical Practice*.

Enthesitis-related arthritis is that which occurs in conjunction with enthesitis. Enthesitis is a separate inflammatory condition where the entheses, the points where the tendons and ligaments attach to the bones, are inflamed, swollen and painful. This is the only type of JIA that is more often seen in boys than girls and it is usually diagnosed after the age of six years. Enthesitis has much evidence pointing towards a genetic predisposition, and a gene (HLA-B27) has been found to be an indication of this condition. The entheses require a very specific type of examination by a physician, but because this type of examination is not known among physicians very well nor is it taught in medical schools, this type of condition is probably very under-diagnosed.

Polyarticular rheumatoid factor (RF) negative JIA and Polyarticular rheumatoid factor positive JIA are two more forms of the condition. The RF negative form is diagnosed when a child has five or more affected joints in their first six months of disease but they do not have a rheumatoid factor. A rheumatoid factor is

a very specific antibody marker in the blood. If this marker is present in at least three blood tests in the first six months of arthritis that affects five or more joints in a child, it is then polyarticular rheumatoid factor positive JIA. The RF negative form usually has more favorable outcomes, with, typically after many years of medication, around 50% of children going into long term remission. RF Positive JIA, on the other hand, usually lasts much longer and is very persistent. This form of arthritis typically continues on into adulthood and those who are diagnosed may be on medication to treat their symptoms for decades. Not only that, but it also can cause joint deformity and significant lack of function if the child does not respond well to treatment.

Systemic JIA, which is the type of arthritis that Mia Nieto was diagnosed with, presents with high spiking fevers and rashes and accounts for about 10% of diagnosed cases of JIA. There is no rheumatoid factor present in blood tests when the arthritis is systemic, and this type typically affects much more than just the joints. Organs can also be affected in this kind of JIA, as evidence by the fact that in Mia's case, there was liver and lymph node involvement and inflammation. This form of JIA tends to last into adulthood as well and can cause significant damage to joints. The highest rates of joint replacements later in life are from children diagnosed with this form of arthritis. However, many children experience their systemic arthritis going into remission if they respond well to medication.

The final form of JIA is known as undifferentiated. This is the diagnosis when the symptoms either do not fit the criteria for any of the other forms of arthritis or, more commonly, the symptoms fit the criteria for multiple kinds of JIA.

These seven different kinds of JIA are essentially what separate juvenile arthritis from adult arthritis. In adults, only one kind of arthritis is typically seen; Polyarticular rheumatoid factor positive. Not only do adults not experience these different kinds of arthritis, the actual arthritis can present very differently. For example, uveitis, which is a major concern in children with arthritis, does not occur in adults.

While there is no known cure for JIA, there are very effective treatment options for the symptoms of the condition. Unfortunately, each child is completely different. Many pediatric rheumatologists describe using different medications in sort of a “guessing game.” There are very typical, very effective routes such as prescribing methotrexate injections and steroid injections and NSAIDS, but when it comes to the specific types of biologic infusions, NSAIDS, etc., each patient is different. Some patients will respond very well to a particular medication while others will not. Not only that, but a child can respond very well to a certain medication and then, all of a sudden, it will stop working and the rheumatologist will have to try a different drug in order to keep the child functioning at as high a level as possible and keep destruction of the body as far away as possible. In Mia’s case, for example, she was doing well on the biologic infusion of Humira when she was 5 and a half. Two year later, however, Humira no longer worked and she was

put on Actemra in order to manage her symptoms. Overall, the medications that treat the symptoms of arthritis are very effective and many children go into remission and are relatively free from swelling, pain, and bodily damage. Of course, it is important to note that in order to treat the symptoms, a child must first receive a proper diagnosis and have the correct medications prescribed.

Perhaps the most problematic issue regarding juvenile arthritis is the fact that there is a serious lack of awareness in the medical community. This problem begins in medical school. Because medical school is designed to teach future doctors of all specialties, the curriculum is extremely focused and comprehensive. However, medical students rarely get exposure to the inner workings of pediatric rheumatology in their studies. In terms of the future practice the subspecialty, less than 1% of all graduating medical students go into a career in the field of rheumatology, both adult and pediatric combined. In the United States, there are only an estimated 250 to 300 practicing pediatric rheumatologists. In terms of diagnostic testing and examination principles, medical students are rarely taught how to diagnose conditions such as juvenile arthritis. Not only that, but they typically are not aware of basic symptoms or how to properly examine joints. As a result, frontline physicians (typically a pediatrician or family doctor) will incorrectly diagnose a child with JIA as having an infection such as Lyme disease or will suspect injury and send the child to an orthopedic specialist. While these children are shipped from doctor to doctor with no diagnosis, their immune systems can do irreversible damage to their bodies.

When it comes to medical education regarding pediatric rheumatology, if a medical student learns about the field in medical school, it is “sheer luck,” says Dr. Joan Von Feldt, a professor of medicine at the University of Pennsylvania and director of the graduate and undergraduate medical education at Philadelphia Veterans Medical Center. Dr. Von Feldt, along with her administrative duties and active practice of adult rheumatology, teaches an introductory course at the medical school in differential diagnosis. In that course, Dr. Von Feldt ensures that every student at the University of Pennsylvania Medical School sees a case study of a child with arthritis. However, what that means is that those students, though they are exposed to pediatric rheumatology, learn about childhood arthritis through required curriculum for only one hour---and---a---half lecture during their entire four---year long medical education. Any other exposure they have is not required or planned in their curriculum. “At the University of Pennsylvania, we’re trying to evaluate the musculoskeletal curriculum and trying to figure out where the deficits are...the clinical skills of performing musculoskeletal exams is poor, so improving the skills of students is a priority,” says Dr. Von Feldt.

Though Dr. Von Feldt teaches only one lecture that is focused on the topic of childhood arthritis at the University of Pennsylvania, she is doing more than many medical schools are. In a 2004 report by the American Academy of Pediatrics, it was found that a third of the nation’s medical schools did not have a pediatric rheumatologist associated with them. Even more worrisome is the fact that in a 2001 survey conducted by Drs. Gary L. Freed, Sandra Jee, Laura Spere and Sarah J.

Clark found that, in a national survey, only “18% of pediatricians and 12% of family practitioners believed they were adequately trained to diagnose and treat juvenile arthritis.”

Medical education is assumed to be better and more comprehensive as time goes on and as new research is conducted and new cures are found. Regarding pediatric rheumatology, however, it seems as though medical education may have actually decreased. This is not a curriculum change that any medical school board made; pediatric rheumatology has never had an important presence in medical schools. It may, in fact, be rather a matter of cost. The Arthritis Foundation, a non-profit organization that aims to raise awareness of the prevalence of arthritis and encourage research in the field, used to deliver a book to medical schools called the *Primer on the Rheumatic Diseases*. This book was handed out to medical students for free, and was first published in 1934. Now in its thirteenth edition, the book is still being published, but instead of being handed out to medical students to raise awareness of rheumatic illnesses, the book is on sale from the Arthritis Foundation for \$79.95. Medical students today must first become aware that the book exists and then buy the book themselves if they wish to gain knowledge of rheumatic illnesses addressed in it. This is understandable, given the current state of the economic situation in the United States, but still represents a massive decrease in the education of medical students. The Arthritis Foundation is in no way required to hand out the book and the publisher absolutely deserves payment, but it nonetheless shows that the little education students once received concerning

pediatric rheumatology is no longer readily available. For medical students of the past, this book may have been the only place they read about rheumatic illness. Now, they must pay a considerable sum of money to read about a branch of medicine they may not even be aware exists.

Because medical schools are lacking in curriculum addressing pediatric rheumatology, doctors tend to get more exposure to the field during the next step in training after graduating from medical school, their residencies, reports Dr. David Sherry, section chief of pediatric rheumatology and full professor at Children's Hospital of Philadelphia, a hospital associated with the University of Pennsylvania. However, even during residency, it is a "hit or miss" system. "If a resident happens to apply to a hospital program where there is a pediatric rheumatology ward, they will see childhood rheumatic cases. If their hospital does not have a pediatric rheumatology department, it is rare that they will ever get exposed to children with rheumatic illness. It's pure luck," says Dr. Sherry.

Issues with getting a correct diagnosis early are seemingly connected with this lack of education in medical schools. Doctors are not trained to look for symptoms of arthritis; they are trained to look for symptoms of more everyday---type problems, such as infection. Dr. Sherry estimates that the average time it takes for a child to get a correct diagnosis of juvenile arthritis is six months after the initial symptoms appear. "The worst that can happen is that these kids go blind," Dr. Sherry says. "Second worse is that they can have deformity of joints, overgrowth of bone, or leg---length discrepancies." These problems, once they arise, can be very

hard to correct. “We have fairly effective treatment” Dr. Sherry says, “but we need to shut the door before the cows get out of the barn, so to speak.” If children are not getting correct diagnoses and being sent to pediatric rheumatologist specialists who can administer correct treatments, those children may have severe problems that can’t be corrected, such as blindness or joint deterioration.

Lack of sufficient medical education is not the only problem when it comes to diagnosing juvenile arthritis. Infections, blood disorders, and other rheumatic illnesses can cause the same kinds of symptoms as arthritis. Therefore, symptoms such as swollen joints, lack of range in motion of joints, and limping are commonly assumed to be the result of something else, says Dr. Sherry. Oftentimes, it *is* something else that causes joint swelling; it’s not always arthritis. Many doctors incorrectly assume that those symptoms are caused by trauma but, in small children, trauma is rarely the cause of those kinds of problems in joints. In order for a diagnosis of arthritis to be made, a symptom, such as the swelling of a joint, must be present for six weeks. If joints are affected for less time, the cause is usually either another condition or perhaps infection. When correct diagnoses are swiftly made, the outcomes are relatively positive for children with arthritis.

Because juvenile arthritis is not nearly as rare as people generally assume, great strides are being made to raise awareness. Organizations such as the Arthritis Foundation, Children and Arthritis, Inc., Childhood Arthritis and Rheumatology Research Alliance, and the American Juvenile Arthritis Organization promote awareness through races, fundraisers, and camps and many raise funds for research

into the potential causes and effective long-term treatments of juvenile arthritis. Several organizations host camps for families who have a member with juvenile arthritis; they can get together and children who have arthritis can meet each other in a non-medical setting. Oftentimes, doctors attend these kinds of get-together camps too. Dr. Ruy Carrasco goes to such a camp, Jambalaya Jubilee, every year. There, he can meet with parents who have a child or children with arthritis and answer questions they may have about the condition, treatment options, etc. These camps oftentimes provide comfortable places for parents and kids to learn more about arthritis from both other families and doctors. However, even though these strides are being made, more awareness is always needed among those who do not have or know a child with juvenile arthritis.

Awareness, medical advances regarding effective medical treatment, and the research being done to find better treatments and perhaps causes and cures of arthritis are some of the ways that the interest and study of juvenile arthritis have advanced over the years. Unfortunately, one still has to ask: if the initial problem is to be addressed by doctors, how are diagnoses going to be made early enough to ease the discomfort and suffering of thousands of children if learning about pediatric rheumatology in medical school is simply a matter of pure, unadulterated luck?